Disorder	Gene	Mutations	Notes	Source
Ashkenazi Jewish Panel: Bloom Syndrome, Canavan Disease, Familial Dysautonomia, Fanconi Anemia Type C, Gaucher Disease, Mucolipidosis Type IV, Niemann-Pick Disease, Tay Sachs Disease	BLM ASPA IKBKAP FANCC GBA MCOLN1 SMPD1 HEXA	31 mutations associated with disorders	AJP Control™ (Ashkenazi Jewish Panel) for the Luminex (TM Bioscience Tag-It™) platform.	Molecular Controls
cystic fibrosis	CFTR	38 CFTR mutations including ACMG 23 panel	Mutations, 3 polymorphisms, and 5/7/9T variants. (in blood-like matrix) FDA cleared	Maine Molecular Quality Controls, Inc.
cystic fibrosis	CFTR	23 CFTR mutations recommended by the ACMG.	Also contains variants: I506V, I507V, F508C, 5/7/9/11T and I148T. (in blood-like matrix) FDA cleared	Maine Molecular Quality Controls, Inc.
cystic fibrosis	CFTR	79 CFTR mutations including ACMG 23 panel	Also contains variants: I506V, I507V, F508C, 5/7/9/11T and I148T. (in blood-like matrix)	Maine Molecular Quality Controls, Inc.
cystic fibrosis	CFTR	32 mutations in the Abbott/Celera Cystic Fibrosis Genotyping Assay. Not available in Canada or EU		Seracare
cystic fibrosis	CFTR	47 mutations- Not available in US or Canada	When used in conjunction with Accurun 676, covers all 81 mutations in the Luminex xTag™ CFTR IVD Assay	Seracare

cystic fibrosis	CFTR	34 mutations. Not available for sale in Canada or EU	When used in conjunction with Accurun 644, covers all 81 mutations in the Luminex xTag™ CFTR 70+6 Assay	Seracare
Hemochromatosis	HFE	H63D, C282Y	Control to monitor extraction, amplification and detection. Mutations are heterozygous, homozygous. Wild type alleles are also included. DNA in blood-like matrix.	Maine Molecular Quality Controls, Inc.
Hemochromatosis	HFE	H63D, S65C, C282Y	Control to monitor extraction, amplification and detection. Mutations are heterozygous, homozygous. Wild type alleles are also included. DNA in blood-like matrix.	Maine Molecular Quality Controls, Inc.
Hemochromatosis	HFE	Wild Type	Control to monitor extraction, amplification and detection. Mutations are heterozygous, homozygous. Wild type alleles are also included. DNA in blood-like matrix.	Maine Molecular Quality Controls, Inc.
Heteroplasmic Mitochondrial DNA Mutation Detection Standard	mtDNA	human mitochondrial DNA mixtures which simulate different levels of heteroplasmy.	mtDNA mixtures (mass % polymorphic levels are 1 %, 2.5 %, 5 %, 10 %, 20 %, 30 %, 40 % and 50 %)	National Institute of Standards and Technology

Huntington	HTT	CAG repeat lengths of 6 samples: 15/29, 17/36, 15/40, 35/45/ 39/50, 17/75	6 genomic DNA samples, 2 alleles per sample. Repeat lengths classifications: normal, intermediate, reduce penetrance, full penetrance	National Institute of Standards and Technology
Mitochondrial DNA Sequencing		Human cell line DNA-sequence of mtDNA is well characterized	Contains DNA extracted from two cell lines plus cloned DNA from a region that is difficult to sequence.	National Institute of Standards and Technology
Pharmacogenetics	CYP2C9, CYP4Fs, VKORC1	CYP2C9: *2, *., *5, *6, *11, *14, *16. VKORC1 -1639G>A, CYP4F2 1347G>A	analytical performance of the extraction, amplification and detection steps performed according to the eSensor® Warfarin Sensitivity Plus Test on the eSensor® XT-8	Maine Molecular Quality Controls, Inc.
Pharmacogenetics	CYP2C19	CYP2C19: *2, *3, *4, *5, *6, *7,, *8, *9, *10, *17	control to monitor analytical performance of the extraction, amplification and detection steps of Cytochrome P450 2C19 (CYP2C19) mutations .	Maine Molecular Quality Controls, Inc.
Pharmacogenetics	CYP2C9, VKORC1	CYP2C9: *2, *3, *4, *5, *6. VKORC1: -1639G>A, 85G>T, 121G>T, 134T>C, 172A>G, 1331G>A, 3487T>G	Synthetic material, mixture of mutations in CYP2C9 and VKORC1 in one tube.	Molecular Controls

Pharmacogenetics	CYP2C19	CYP2C19: *2, *3, *4, *5 (A,B), *6, *7, *8	Synthetic reference nucleic acids comprising 7 mutations/variants in the cytochrome p450 CYP2C19 gene.	Molecular Controls
Pharmacogenetics	CYP2D6	CYP2D6: *2A promoter, *2 (1661G>C), *2 (4180G>C), *2/*17, *3 (A,B), *4(A-L), *4 (A-L)/*10 (A,B), *5 deletion, *6 (A-D), *7, *8, *9, *12, *11, *14, *15, *17, *41	Synthetic reference nucleic acids comprising 17 mutations/variants in the cytochrome p450 CYP2D6 gene and the *5 deletion.	Molecular Controls
Thrombosis	Factor II, Factor V	Factor II G20210A, Factor V Leiden G1691A	Intended for in vitro use as a quality control to monitor analytical performance of the Xpert HemosIL® Assay on the GeneXpert® System. Mutations are homogenous, wild type included also.	Maine Molecular Quality Controls, Inc.
Thrombosis	Factor II, Factor V	Factor II G20210A, Factor V Leiden G1691A	Mutations are heterozygous. Wild type alleles are also included.	Maine Molecular Quality Controls, Inc.
Thrombosis	Factor II, Factor V	Factor II G20210A, Factor V Leiden G1691A	Mutations are heterozygous, homozygous. Wild type alleles are also included.	Maine Molecular Quality Controls, Inc.
Thrombosis	Factor II, Factor V	Factor II G20210A, Factor V Leiden G1691A	Control to monitor extraction, amplification and detection. Mutations are heterozygous, homozygous. Wild type	Maine Molecular Quality Controls, Inc.

Thrombosis	MTHFR	C677T, A1298C	contains: 1) wt MTHFR; 2) heterozygous 677C>T and 1298A>C genotypes; 3) homozygous 677C>T and 1298A>C genotypes (in blood-like matrix)	Maine Molecular Quality Controls, Inc.
Thrombosis	factor II, Factor V, MTHFR	Factor II G20210A, Factor V Leiden G1691A, MTHFR C677T, A1298C	Control to monitor extraction, amplification and detection. Mutations are heterozygous, homozygous. Wild type alleles are also included.	Maine Molecular Quality Controls, Inc.

Product	catalog #
AJP Control	https://www.molecularcontrols.com/Default.aspx?id=1830
INTROL CF Panel I	http://mmqci.com/company.php
INTROL CF Panel II	http://mmqci.com/company.php
INTROL CF Panel III	http://mmqci.com/company.php
Accurun 632	http://www.seracarecatalog.com/Default.aspx?tabid=445
Accurun 644	http://www.seracarecatalog.com/Default.aspx?tabid=445

Accurun 676	http://www.seracarecatalog.com/Default.aspx?tabid=445
INTROL HH Panel I Control	http://mmqci.com/company.php
INTROL HH Panel II Control	http://mmqci.com/company.php
INTROL HH Wild Type Control	http://mmqci.com/company.php
SRM2394	http://www.nist.gov/index.html

SRM2393	http://www.nist.gov/index.html
SRM 2392	http://www.nist.gov/index.html
INTROL PGx 1 Control	http://mmqci.com/company.php
INTROL 2C19 Panel P105	http://mmqci.com/company.php
P450-2C9 VKORC-1 Synthetic Control	https://www.molecularcontrols.com/Default.aspx?id=1832

P450-2C19 Synthetic Control	https://www.molecularcontrols.com/Default.aspx?id=1832
P450-2D6 Synthetic Control	https://www.molecularcontrols.com/Default.aspx?id=1832
Xpert FII & FV NOR/MUT Control	http://mmqci.com/company.php
Xpert FII & FV HET	http://mmqci.com/company.php
Xpert FII & FV Genotype Panel G109	http://mmqci.com/company.php
INTROL TRC Genotype Control	http://mmqci.com/company.php

INTROL MTHFR Genotype Control	http://mmqci.com/company.php
INTROL Thrombosis Genotype Panel	http://mmqci.com/company.php